

Patient Name: Sample Patient (fetus of)

DOB: Age:
SSN #: Gender: Female

City Hospital
1 Main Street
Anywhere, USA

Genzyme Specimen #: 10000001

Case #: Patient ID #: 20000002
Date Collected: 04/08/2009 Date Received: 04/08/2009

Referring Physician: John Doe, MD

Genetic Counselor:

Client Lab ID #:

Hospital ID #:

Specimen ID #:

Specimen(s) Received: 2 - Flask(s)

Specimen Type: Cultured AF cells

Ethnicity: Ashkenazi Jewish

Clinical Data: Prenatal Test/Family History - both parents carriers

RESULTS: SMN1 copy number: 0 (See Interpretation)

INTERPRETATION:

This fetus has no copies of SMN1 and is therefore predicted to be affected with spinal muscular atrophy, a disease of variable age of onset and severity. Genetic counseling is recommended.

COMMENT:

Comparison of maternal and fetal DNA markers indicates that maternal cell contamination is unlikely to have interfered with the reported fetal result (maternal specimen #12345678).

Spinal muscular atrophy (SMA) is an autosomal recessive disease of variable age of onset and severity caused by mutations, most often deletions or gene conversions, in the survival motor neuron (SMN1) gene. Among individuals with a clinical diagnosis of SMA, ~94% lack both copies of the SMN1 gene (i.e. 0 copies of SMN1), while other affected individuals may have both a deletion and a non-deletion mutation. Additionally, de novo mutations have been reported in ~2% of SMA patients.

This dosage analysis cannot detect: 1) non-deletion mutations within the SMN1 gene; 2) germline mosaicism; or 3) mutations in genes other than SMN1.

METHOD/LIMITATIONS:

Specimen DNA is isolated and amplified by real-time polymerase chain reaction (PCR) for exon 7 of the SMN1 gene and two reference genes. A mathematical algorithm is used to calculate the number of copies of SMN1. Sequencing of the primer and probe binding sites for the SMN1 real-time PCR reaction is performed on all fetal samples, and on samples from individuals with one copy of SMN1 on carrier testing, to rule out the presence of sequence variants which could interfere with analysis and interpretation. False positive or negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation, erroneous representation of family relationships or contamination of a fetal sample with maternal cells.

REFERENCES:

1. Smith M, Calabro V, Chong B, et al. 2007. Eur J Hum Genet 15:759-766. 2. Online review of SMA: <http://www.genereviews.org/profiles/sma>

The test was developed and its performance characteristics have been determined by Genzyme. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing. This test must be used in conjunction with clinical assessment, when available.

Electronically Signed by: D. Alexa Sirko-Osadsa, Ph.D., FACMG, on 4/15/09

Reported by: ES/es

Testing performed at Genzyme Genetics 3400 Computer Drive, Westborough, MA 01581 1-800-255-7357

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